

## Delivering improved care for families with inherited cardiovascular conditions

Inherited cardiovascular conditions (ICCs) are a group of genetic disorders that affect the heart and blood vessels; this includes some disorders that also affect other body systems. Sometimes, the first evidence of an ICC in a family is the sudden and unexpected death of an apparently healthy young person; other relatives may also be at risk. Recent advances in genetics and cardiology are now making it possible for doctors to offer more accurate diagnosis, improved clinical management, and better risk assessment for families affected by ICCs - avoiding tragic and unnecessary deaths. Steps are now needed to ensure that speedy and equitable access to these services is possible across the UK.

A new report from the Foundation for Genomics and Population Health (PHG Foundation), *Heart to Heart: Inherited Cardiovascular Conditions Services*, provides a needs assessment and overview of current service provision for patients and families affected by inherited cardiovascular conditions in the UK.

This work was funded and led by the PHG Foundation, and carried out in conjunction with an external Working Group of experts including NHS cardiologists, geneticists, service commissioners and managers and representatives from patient groups and charities the British Heart Foundation (BHF), Cardiac Risk in the Young (CRY), the Cardiomyopathy Association, the Marfan Association UK and Sudden Arrhythmic Death Syndromes (SADS) UK.

A survey of NHS service providers showed that capacity is insufficient to meet current or anticipated future needs for ICCs in the UK, and that there are serious inequalities in the quality and quantity of services available to patients in different parts of the country. The report sets out the requirements for appropriate specialised services for ICCs, which should include multidisciplinary teams of health professionals, access to the latest forms of testing and clinical investigations and provision of bereavement support and counselling. Increased awareness of these integrated services (for example, among GPs) and co-ordination with voluntary support organisations are essential.

The Working Group concluded that every UK cardiac network should ensure that its population has access to specialised expert ICC services for children and adults, although most will not have their own service. Other key recommendations included:

- Cardiac networks and experts should develop agreed minimum standards for provision of and access to specialist ICC services, according to appropriate timeframes and taking into account anticipated increases in demand over the next 5-10 years.
- ICC service providers should work with others to ensure that the necessary support systems and a workforce with the necessary specialist competences are developed, and that suitable research and evaluation keeps services up to date.
- Legislative and system changes should be sought to ensure appropriate retention and handling of tissue samples following sudden cardiac death. Best practice in terms of consent, data sharing and clinical testing should be promoted, and the responsibility of coroners to family members who may be at risk of sudden cardiac death clarified.

PHG Foundation Programme Director and report co-author Dr Hilary Burton commented: *"Our report provides a starting point for the development of high quality ICC services across the UK, but there is much more to do. Progress will require leadership and a great deal of cooperation and commitment from commissioners, service providers and patients"*.

National Clinical Director for Heart Disease and Stroke, Professor Roger Boyle said: *"The Department of Health is committed to reducing the number of sudden cardiac deaths as well as improving services for victims' families. In October 2008, we launched a new database for pathologists to record sudden cardiac death and improve our understanding of this inherited heart condition. The database is helping to identify people who are unknowingly at risk so that we can refer them to*

*specialist centres where they will be offered counselling and helped to reduce their risk of sudden cardiac death. We welcome this report and will be working closely with the NHS, local healthcare services and charities, including PHG Foundation, to develop these specialist inherited cardiac services and address issues surrounding access".*

**Professor Paul Brennan, Consultant in Clinical genetics at Teeside Genetics Unit and a member of the expert Working Group said:** *"Together, inherited cardiac conditions are a common cause of preventable ill-health and sudden death, often in young people. The PHG Foundation's report provides an invaluable and timely survey of current clinical and laboratory services for these people and their families...In short, despite apparent current deficits in service provision, the NHS is capable of providing the most comprehensive, equitable, patient-centred network of specialist services for inherited cardiac conditions in the world. The clinical and laboratory genetics communities in the UK welcome this report as a foundation for future collaborative service development".*

**Another member of the Working Group, Professor Hugh Watkins (Field Marshall Alexander Professor of Cardiovascular Medicine, Oxford University) commented:** *"Inherited cardiovascular conditions, while not as common as heart disease in later life, do result in a particularly severe burden for affected families. Fortunately this is an area where genetic research has led to real advances in diagnosis and treatment but, because the conditions are not common, the best care can really only be provided by highly specialised services. So the lead that the PHG Foundation has taken to bring all the interested parties together, and to prepare a consensus report on the components needed for optimal ICC services, is timely and of lasting value".*

**Alison Cox, founder and chief executive of the charity Cardiac Risk in the Young (CRY), commented:** *"These tragedies with their genetic implications are catastrophic for families. Grief is exacerbated if the young person has had symptoms that have not been recognised and treated resulting in a sudden death that was avoidable. Fast-track clinical expertise is crucial. This ambitious and comprehensive report by the PHG Foundation provides a vital first step in recognising the complicated and challenging issues involved. The needs of the patients should always be paramount and the sensitivities involved in dealing with those affected have been recognised in this document. An enormous amount of time has been invested in compiling this wide-ranging review that highlights how far we have yet to go before equitable service provision is established".*

The full report and a shortened summary version (including the main recommendations) are available from 15<sup>th</sup> June 2009 from the PHG Foundation website, [www.phgfoundation.org](http://www.phgfoundation.org)

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#### Notes to Editors

- The PHG Foundation is an independent, not-for-profit, multidisciplinary health policy research and development organisation based in Cambridge, UK, but working internationally.
- We focus on 'bridging the gap' in clinical translation to move new genomic and biomedical knowledge and applications into health service practice and better health for people and populations.
- We are the leading UK centre for public health genomics, and have considerable experience in policy research and development, as well as evidence-based evaluation of clinical tools and health services.
- Our expertise in areas including the scientific, clinical, public health, ethical, legal, social and regulatory aspects of genomics and biomedicine.

*The PHG Foundation is the working name of the Foundation for Genomics and Population Health, a charitable company registered in England and Wales, charity no. 1118664 / company no. 5823194.*

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